Molecular analysis, Clinical Manifestations and Management of Factor XIII Deficiency in Southeast of Iran

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BACKGROUND

Factor XIII deficiency is a rare bleeding disorder(RBD) that has an extremely high incidence in Sistan and Baluchistan Province located in southeast of Iran. This disorder presents with various clinical manifestations ranging from mild to life threatening symptoms. Due to high prevalence of this disorder in this province, the aim of our study was to evaluate molecular analysis, clinical features and management of this disease.

CONCLUSION

Trp187Arg is the most common mutation of FXIII-A subunit in southeast of Iran and this area has the highest prevalenceof factor XIII deficiency in the world, and prophylactic treatment waseffective in management of disease.

METHODS

This study was conducted on 190patients with severe factor XIII deficiency. Initially ,suspected patients were diagnosed base on previously reported mutation of Trp 187 Arg of subunit A of FXIII gene. An informed consent was signed by each participant. The medical records and data of each individual including demographic characteristic, clinical manifestations and treatment of disease were extracted. Eventually, the obtained data of patients was analysis by SPSS software.

RESULTS

Molecular analysis of this large group of patients with severe factor XIII deficiency revealed that all studied patients were homozygous for TGG CGG mutation at codon 187, in exon 4 of FXIII-A1 gene. Among190 patients, 100 (52.6%) were male and 90 (47.4%) were femaleand the mean age of them was 26±3 years.The most common symptom in affected individuals is bleeding from umbilical stump (83%) that observed few days after birth and is a diagnostic manifestation in FXIII deficiency. The other frequent symptoms were deep soft tissue (52%) hematoma and *Prolonged wound Bleeding*(30%) while the rarest one was prolonged menstrual bleeding (0.5%). CNS bleeding as a debilitating manifestation was reported in 50 patients. Prophylactic treatment including cryoprecipitate, FFP and Fibrogammin Pin dose of one bag per 10 – 20 kg every 3 – 4weeks, 10 mL/kg every 4 – 6 weeks and 10 to 35 IU/kg every 4 – 6 weeks respectively, were successful in preventing of CNS bleeding, miscarriage and even minor bleeding episodes.

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